XP Summer News

NIGHTTIMES

A newsletter from the Xeroderma Pigmentosum Family Support Group, a registered non-profit organization dedicated to helping those touched by the rare genetic condition through education, research and kindness.

Newsletter

Peyton's Story

Peyton Madden is a typical 10 year old. He loves Star Wars, Pokémon, riding his bike and playing with Legos. If you meet him inside, you would never know that he is unique other than an abundance of freckles and twinkling eyes that speak of mischief. However, if you meet him outside, you will instantly notice a difference. Peyton was diagnosed with Xeroderma Pigmentosum (XP) when he was three years old. That meant he couldn't repair the damage from ultraviolet light and had to be covered head to toe whenever he went outside. His parents still remember the day the dermatologist suggested that he had XP and the sense of loss experienced by those living with a rare, incurable disease. "You mourn the life that could have been," says his mother, Sarah, "I remember sitting on the laundry room floor with Peyton's dad, Kyle as we discussed how we would adjust to this new life." Like most families, the thought of what was needed to provide a safe life for Peyton was overwhelming. On-line research led Sarah to the XP Family Support Group's website. "There were two groups offering help to families with XP," says Sarah, "I chose to reach out to the XP Family Support Group because their website was full of ways to help families and kids with XP." Thanks to the XP Family Support Group, families learn how to live life to the fullest and how to not let XP hold them back. Seven years later, Peyton has visited the Bahamas, Mexico, Belize and Honduras. He's gone on a Disney Cruise, climbed a Mayan ruin, hiked in Yellowstone and sat in the dugout during batting practice with the 2015 World Champion Kansas City Royals. Now his mom says, "He's just a normal little boy living a BIG life!"

Want to see your child featured here? Submit your article to Newsletter@xpfamilysupport.org

Research Update

By Laura Niedernhofer

Dr. Laura Niedernhofer is a researcher at Scripps Florida. Her lab is testing novel therapeutics in DNA repair deficient mice with the hope of finding treatments for rare diseases like XP as well as degenerative diseases of old age.

Do you guys remember Tania Rozgaja from the 2014 Kansas City XPFSG retreat? She developed a way to measure nucleotide excision repair in human blood cells. Basically, you isolate cells from blood, zap them with UV light, then measure how much new DNA the cells make in response to the UV. Any new DNA detected specifically means the cells did nucleotide excision repair (NER) of UV damage. The test is called measuring unscheduled DNA synthesis (UDS) and it is used to diagnose XP.

The UDS assay has been around for decades. But Tania made the assay better by adapting it from using skin cells to blood cells. So now the test can be done in 24 hours instead of waiting 3 months for skin cells to grow.

Tania has had a great couple of years. She got married to her longtime boyfriend Chris. She got a great job in Texas organizing clinical trials. And she is now expecting her first child, a boy!

Amira and Matt in my lab are continuing to optimize the assay. Currently, we are using the assay to test cells from a young boy in Mexico who is aging very quickly. His doctors suspect that he has "progeria" due to a mutation in *XPF*.

The Niedernhofer lab also received a contract from the National Institute of Health (NIH) Undiagnosed Disease Program (UDP). This is a program in Bethesda, MD where patients with very rare diseases try to figure out what they have. The UDP identified several patients who start to develop neurodegeneration in their 30's and 40's. They suspect that the patients have a mutation in an XP gene. If this is the case, the patient will have low UDS and we can help the doctors with the diagnosis.

Next, we hope to offer this test to any of you who are interested. The results will tell you what %UDS you have, near normal, half-way, or very little.

Amira is headed to medical school at NYU next fall. She plans to be dermatologist.

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The Nobel Prize in Chemistry 2015

By Laura Niedernhofer

2015 was a big year for DNA repair! Finally, the most prestigious scientific award in the world was awarded for the discovery of DNA repair mechanisms. This seals the deal that DNA repair is vitally important – and hopefully more people will hear about it.

Photo: A. Mahmoud

Tomas Lindahl

Prize share: 1/3

Photo: A. Mahmoud

Paul Modrich

Prize share: 1/3

Photo: A. Mahmoud

Aziz Sancar

Prize share: 1/3

The Nobel Prize in Chemistry 2015 was awarded jointly to Tomas Lindahl, Paul Modrich and Aziz Sancar *"for mechanistic studies of DNA repair"*.

Each of the three Nobel laureates discovered a different DNA repair mechanism.

Tomas Lindahl discovered base excision repair (BER). This is the pathway that removes DNA damage caused by normal metabolism. This damage is what we call endogenous DNA damage, meaning it happens inside of us, and it is next to impossible to avoid. Absence of base excision repair is incompatible with human life.

Paul Modrich discovered mismatch repair (MMR). This is the pathway that removes errors made when DNA is copied (a process required to make a new cell). If a person is missing mismatch repair, they have Lynch syndrome (also called hereditary nonpolyposis colorectal cancer HNPCC) and have virtually a 100% chance of developing colon cancer.

Aziz Sancar discovered nucleotide excision repair (NER). This is the pathway that protects us from DNA damage caused by UV light. It also protects us from endogenous DNA damage that is too big for BER to handle. As you well know, if a person has a defect in NER, they have xeroderma pigmentosum (XP), Cockayne syndrome (CS) or trichothiodystrophy TTD.

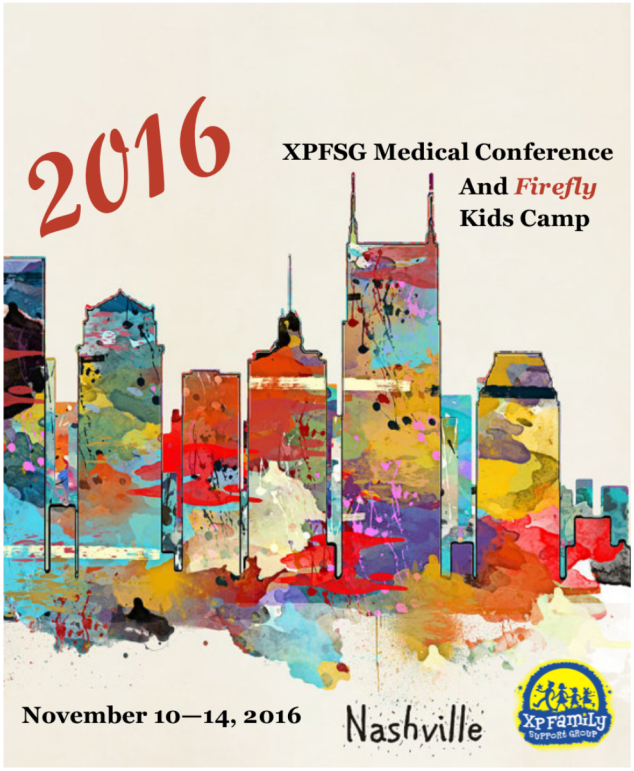
The Nobel Prize for the discovery of DNA repair mechanisms means that the world's top scientists recognize that the repair of damaged DNA is of extraordinary importance. Perhaps this means that more people now understand what DNA and its repair are and why they are important. Hopefully, they will also be more understanding about how hard it is to live without it.

It is no surprise to me that a Nobel Prize was given for DNA repair. All of us (me and all of you) knew DNA repair was vital long before the Royal Swedish Academy of Sciences (who give out the Nobel prize) figured it out. But what surprises me is that the Prize was in Chemistry and not Physiology or Medicine. If the award had been given in Physiology or Medicine, the attention would not be on the mechanism of repair but on how DNA repair impacts our health? The answer to this question is everything XP patients have taught us about the danger of the sun, the link between sunburn and skin cancer, and that DNA damage can impact hearing and nerve function. Perhaps if the prize was in Physiology and Medicine, some of our heroes like Jim Cleaveland and Phil Hanawalt, would have won the prize. Because they helped make the first links between DNA damage, repair and XP.

So I feel a little cheated for the lost opportunity to shine light on XP and remind folks that you have taught us so much about the causes of cancer, aging and neurodegeneration. But a Nobel Prize in Chemistry still helps highlight the importance of DNA repair. And if we don't understand the mechanisms of DNA repair, we will never find a cure for XP.

N I G H T T I M E S

Are you ready for a weekend full of learning and making life-long friendships?

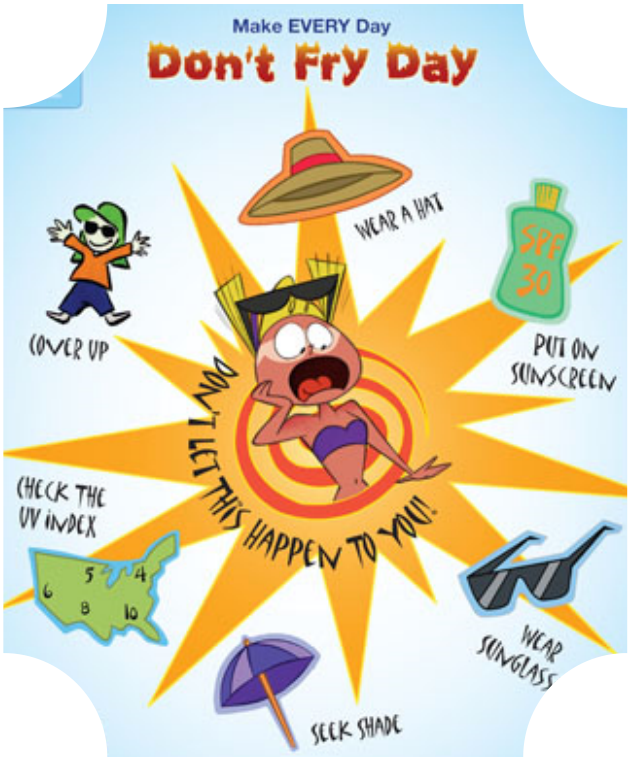


The 2016 XPFSG Medical Conference and Firefly Kids Camp will be held in Nashville, TX November 10—14, 2016. If you have not yet registered to attend, contact our office immediately at www.xpfamilysupport.org. This will be a life changing event for individuals of all ages in the Xeroderma Pigmentosum community. The conferences increase self-esteem, self-image and allow people to discover that they are not alone. There are opportunities for people to interact with others that share the same challenges. Attendees also learn the most current medical research, advocacy and awareness information.

The Best Dietary Sources of Vitamin D

Friday, December 12, 2014 by Michael Ravensthorpe

Vitamin D is one of the most important vitamins needed by our bodies to maintain optimum health. It facilitates calcium absorption in our stomachs, modulates cell growth, regulates our mood and much more. When we are deficient in vitamin D, we often begin to suffer from bone pain, muscle weakness, cardiovascular conditions, low moods and countless other health conditions.



Since human skin cells are able to manufacture vitamin D using certain wavelengths of ultraviolet B light, sunlight will always remain the greatest source of this essential vitamin.

However, those who suffer from Xeroderma Pigmentosum (XP) will have to obtain their vitamin D from food alone (unless willing to take a supplement). Fortunately, a small number of natural foods do contain respectable amounts of the all-important "sunshine vitamin."

Foods rich in vitamin D

- Cod Liver Oil
- Oily Fish
- Mushrooms
- Other Sources– Eggs, Tofu, Raw Milk

Please consult with your doctor to assess your vitamin D levels and prescribed treatment.

Want to add someone to our electronic newsletter? Contact the office at 916-628-3814 or email us at mmilota@xpfamilysupport.org